Genetics Task Force Subcommittee Report

Subcommittee Title: Use of Genetic Information in Health Care
Subcommittee Chair: C. Ron Scott
Subcommittee Members: Robin Bennett, Robert Miyamoto, Maureen Callaghan, Julie Sanford-Hana
Date of Report: June 14, 2002

Part I Diagnosis of Symptomatic Conditions

I. Background
   1. DNA analysis is used routinely in the medical laboratory to identify alterations in genes that are responsible for disease states. It is routine for physicians to request DNA analysis of blood samples from children with mental retardation who are suspected of having the Fragile-X syndrome, from males with symptoms of Duchenne muscular dystrophy, from persons with a clotting disorder, or from adults with muscle and neurologic changes suggestive of a genetic condition.
   2. The introduction of DNA testing has simplified the medical diagnosis of these and other conditions that in the past may have involved anesthesia, muscle biopsies, or expensive and laborious testing by other means.
   3. The committee believes that the use of DNA testing for medical diagnosis of symptomatic individuals is appropriate and falls within the general realm of laboratory testing for medical reasons.

II. The incidence of discriminatory actions based upon genetic information
   A. Findings
      1. In reviewing material related to DNA testing for medical diagnostic purposes, the committee could find no examples of discrimination that had occurred by the use of DNA testing.
      2. As heard by the GTF on February 25, the Washington State Human Rights Commission has not received complaints of discrimination resulting from the use or generation of genetic information for diagnostic health care purposes.
      3. Furthermore, the committee finds that DNA testing is an efficient and cost-effective modality for accurately diagnosing genetic disorders.
   B. Conclusions
      1. The committee concludes there is no increased risk of discrimination based on DNA testing for individuals with symptomatic disorders, but rather finds the technology appropriate for medical diagnostic purposes.

III. Strategies to safeguard civil rights and privacy related to genetic information
   A. Findings
      2. The committee finds that the current laws and regulations regarding privacy of medical records are in place and are covered by hospital policy, Washington state statue, and national HIPAA regulations.
      3. Furthermore, individuals symptomatic for a genetic disorder may have protection under the Americans with Disability Act.

B. Conclusions
    1. The committee concludes that information obtained by DNA testing for symptomatic conditions should become part of the medical record, similar to other testing that would be performed for medical diagnosis.

IV. Remedies to compensate individuals for inappropriate use of genetic information
A. Findings
    1. The committee finds that the current legal tort system exists for compensation of individuals for the inappropriate use of medical information.
B. Conclusions
    1. The committee concludes that no additional safeguards are necessary for this category of DNA testing.

V. Incentives for further research and development on the use of DNA to promote public health, safety and welfare
A. Findings
    1. The committee finds that adequate incentives exist within the medical research community to develop DNA testing as an efficient and cost-effective method of diagnosing medical conditions.
B. Conclusions
    1. As the technology improves, DNA testing will also be introduced into the public health system as an adjunct to newborn screening for treatable genetic diseases. This will promote and assist the safety and welfare of young children detected with treatable disorders.
    2. The committee is supportive of this use of DNA testing for the benefit of public health.

<u>Part II Use of Genetic Information for Reproductive Decisions</u>

I. Background
    1. DNA technology is a powerful tool in the arena of reproductive medicine.
    2. In general, two categories of DNA testing exist: (1) identification of pregnant couples at risk for a genetic disease that will cause severe disease in a future newborn; and (2) utilization of DNA technology in pregnancies at high risk for a severe genetic condition.
        a. An example of the first scenario is represented by a recommendation by the American College of Obstetrics and Gynecology that pregnant couples be screened for a battery of mutations that are associated with cystic fibrosis. The identification of a mutation in an asymptomatic pregnant woman would lead to the testing of the father of her child. If both were found to be carriers of a gene for cystic fibrosis, genetic counseling would be offered and prenatal testing of the fetus would be a voluntary option.
        b. The second scenario involves a couple who have previously given birth to a child with a serious genetic condition for which DNA technology can identify whether the current pregnancy is

affected. The couple would be offered DNA testing as a part of genetic counseling to allow them to make a personal reproductive decision. In this situation, DNA testing is appropriate, low risk for mother and fetus, and can accurately distinguish an unaffected from an affected fetus. In this scenario, DNA testing is voluntary on the part of the couples at risk and offers a means for obtaining accurate information at minimal risk and cost, and with a high degree of accuracy.

II. The incidence of discriminatory actions based upon genetic information
   A. Findings
      1. The committee finds that there is little, if any, risk of discrimination based upon the use of DNA technology in the above scenarios.
      2. The testing of couples or fetuses is always voluntary, done with informed consent, and information is maintained in the medical records of the individuals requesting the testing.
   B. Conclusions
      1. The committee concludes there is no need for legislation to expand protection of personal privacy in the area of prenatal DNA testing.

III. Strategies to safeguard civil rights and privacy related to genetic information
   A. Findings
      1. The committee finds that prenatal Genetic information that is contained within hospital or medical records comes under the purview of protection by hospital policy, Washington state statue, and federal HIPAA regulations.
   B. Conclusions
      1. The committee concludes that risk of inappropriate use of the genetic information is the same as for other medical testing performed voluntarily for individuals.
      2. The committee concludes there is no necessity to expand this protection.

IV. Remedies to compensate individuals for inappropriate use of genetic information
   A. Findings
      1. The committee concludes that any breech of confidentiality by the above facilities would lend itself to tort action by the legal profession and censure by the appropriate medical oversight bodies or licensing bureaus of Washington state.
   B. Conclusions
      1. The committee concludes there is no necessity to expand this protection.

VI. Incentives for further research and development on the use of DNA to promote public health, safety and welfare
   A. Findings
      1. The committee finds that active research is being performed within the medical community to expand genetic testing as an aid for reproductive health of mother and fetus.
      2. There exists funding from government and private agencies to expand this field of endeavor.

3. Techniques are being developed that will use extremely small samples of amniotic fluid, maternal blood, or fetal cells to identify DNA alterations that will detect infectious agents or serious genetic conditions.

B. Conclusions

1. The committee concludes there is no need for legislation to protect individual privacy in this particular arena. Adequate safeguards exist within the research community (IRBs), Washington state law, and HIPAA regulations.

Part III Predictive Identification of Genetic Risk Factors for Late-Onset Diseases

I . Background

1. In certain instances, DNA testing can identify genetic predisposition to disease prior to the onset of clinical symptoms.
2. There are two types of situations relevant to this issue.
    a. The first situation occurs in the testing of young children at high risk to develop a serious disorder for which intervention may be available. An example would be a child born into a family in which there exists a previous child diagnosed with cystic fibrosis. The second infant may be asymptomatic, but accurate DNA testing would allow for the identification of that infant as affected or unaffected with cystic fibrosis. If affected, appropriate intervention strategies would begin at the earliest time to help prevent clinical complications. Similar scenarios exist for the recognition of boys born into a family with Duchenne muscular dystrophy, or a young child born into a family at risk for a genetic disease for which there is available therapy. In this case the issues would be the same as those described in the section related to Diagnosis of Symptomatic Conditions.
    b. The second category of predictive testing is more complicated. There exist a number of disorders with clinical symptoms that present in adulthood, but which can be predicted to occur prior to symptoms with a finite probability if an individual carries a particular form of the gene responsible for the disorder. Examples include the predilection for breast cancer in women who carry an abnormality of the BRCA1 or BRCA2 gene, or the predilection for neurological degeneration around the age of 40 in individuals with an abnormality of the Huntington disease gene. DNA technology has the potential to identify individuals at risk for these conditions at any age prior to the onset of symptoms. In the case of a woman with a strong family history of breast cancer, it may be appropriate to screen that woman by DNA testing to determine her

genetic risk to develop breast cancer. Screening would allow for early detection or prevention of breast cancer in a woman with the mutation in BRCA1 or BRCA2. In the case of Huntington disease, an autosomal dominant condition, children of an affected individual are at 50% risk for developing the condition in adulthood, but there exist no medical strategies for treatment or cure. DNA testing is appropriate for medical information and for personal decision-making on lifestyle changes in the case of individuals at risk for Huntington disease.

II. The incidence of discriminatory actions based upon genetic information

A. Findings

1. The committee finds no obvious discrimination documented within the state of Washington based on information obtained by DNA testing on the predictive identification of late-onset disorders.

B. Conclusions

1. It is this category of the use of genetic information, however, that may place individuals at risk for genetic discrimination should such information exceed the bounds of the medical care system. For example, a woman identified in a family with an abnormality of a BRCA1 gene could theoretically be discriminated against in obtaining health insurance or employment because of the perceived increased fiduciary risk she would present to an employer or in social stigmatization. Similarly, an individual identified at age 20 as carrying the gene for Huntington disease could be discriminated against in employment, obtaining health insurance, or from individual or group life insurance.

III. Strategies to safeguard civil rights and privacy related to genetic information

A. Recommendations

1. For individuals tested for presymptomatic genetic conditions, the committee recommends that such information be retained within a separate section of the medical record (similar to psychiatric evaluations). This genetic information would not be transmitted upon routine requests for medical information, but rather would require the <u>specific</u> informed consent of the individual or a court order.

B. Comments

1. (Comment: (1) Persons who are at risk for a late-onset disorder, who determine their genotype, and who find they are at “high risk” for the disorder have an obligation to reveal their risk factor if applying for “supplemental” life insurance. This recommendation is intended to prevent “adverse selection” of high-value life insurance by persons with a predictably shortened life expectancy.
[Supplemental: life insurance over and above group policies or a personal policy obtained subsequent to genetic testing.])

IV. Remedies to compensate individuals for inappropriate use of genetic information
- A. Findings
  1. The inappropriate use of private genetic information for predictive diseases would fall under the recommendations from another portion of the Genetic Task Force Report.

V. Incentives for further research and development on the use of DNA to promote public health, safety and welfare
- A. Findings
  1. The committee finds that incentives for research and development on the use of DNA to promote predictive testing of late-onset diseases is an active research endeavor within the medical community.
  2. There is research and funding available for predicting individuals at risk for developing diabetes, hypertension, renal disease, and cardiovascular disorders for which intervention strategies may be available.
- B. Conclusions
  1. The committee concludes that development of testing for risk factors associated with these common diseases will have a beneficial effect on public health policy and the welfare and safety of the population. The research should be encouraged to continue.

VI. Additional Findings, Conclusions and Recommendations
- A. Findings
  1. The state of Washington has inadequate resources for genetic counseling of individuals seeking or receiving genetic testing.
- B. Recommendations
  1. It is recommended that serious attention be given to establishing a graduate program in genetic counseling at the University of Washington to address this deficit.